SCOLIOSIS SURGERY IN THE PRADER–WILLI SYNDROME

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There is a high incidence of spinal deformity in children with the Prader–Willi syndrome. We have encountered major complications following spinal surgery in this condition. We report our experience and conclude that spinal surgery is a formidable undertaking and the risks should be appreciated by the surgeon and the parents.

The Prader–Willi syndrome (PWS) is an uncommon sporadic disorder occurring in about 1 in 10 000 to 25 000 live births in all races. It is characterised by infantile hypotonia, hypogonadism, early childhood obesity due to hyperphagia, diabetes mellitus, delayed psychomotor development, mental deficiency, short stature, small hands and feet, hypermobile joints, strabismus, myopia, scoliosis, kyphosis and a typical facies with blonde hair, blue almond-shaped eyes, narrow bifrontal diameter and a down-turned mouth. Males outnumber females by three to one (Pearson, Steinbach and Bier 1971; Gurd and Thompson 1981; Holm and Laruen 1981; Butler and Meaney 1987; Cassidy 1987).

PWS is diagnosed on clinical criteria. No absolute diagnostic criteria exist although 50% to 70% of patients have a deletion of chromosome 15 (Cassidy 1987). The prevalence of spinal deformity varies greatly: for scoliosis it is reported to be between 5% and 85% (Laurance 1967; Gabilan and Royer 1968; Zellweger and Schneider 1968; Smith et al 1970; Pearson et al 1971; Clarren and Smith 1977; Gurd and Thompson 1981; Holm and Laruen 1981; Cassidy 1987) and for kyphosis it is between 8% and 40% (Pearson et al 1971; Gurd and Thompson 1981; Holm and Laruen 1981). Holm and Laruen (1981) emphasise that clinical examination alone is not a reliable method to detect spinal deformity in these obese hypotonic patients and suggest that all patients should have spinal radiographs. We know of only four reports of surgery for spinal deformity in PWS, and all the patients had uneventful operations for scoliosis (Gurd and Thompson 1981; Holm and Laruen 1981; Soriano, Weiss and Houghton 1988).

We report our experience of scoliosis surgery in PWS in view of the difficulties we encountered during and after operation.

PATIENTS AND METHODS

Case 1. A girl was diagnosed as having PWS at the age of four years. She was short and grossly obese (Fig. 1) and had been hypotonic from birth (reduced fetal movements had been noted during pregnancy) with...
pronounced ligamentous laxity and greatly delayed milestones. A scoliosis had been noted at the age of 12 months. Despite bracing, the curvature continued to deteriorate and measured 78° from T6 to T12 at the age of five (Fig. 2). She was dyspnoeic at rest with poor exercise tolerance. Her respiratory rate was 80 per minute with a tidal volume of 0.1 l. Despite her poor pulmonary function, corrective surgery was thought to be indicated because of the severity of the curve.

An anterior growth arrest was performed, in which five discs and vertebral end plates were removed from the apex of the curve. All tissues were noted to be friable, and profuse bleeding was encountered, including some from the annulus fibrosus. Rib was used as the bone graft. Intra-operative blood loss was 800 ml and a further 230 ml drained from the chest over the first two hours postoperatively. Recovery was uneventful.

Two weeks later we attempted posterior spinal fusion. Again, profuse bleeding was encountered and the bones proved to be too thin and soft to hold any instrumentation. Therefore the facet joints were excised, the laminae decorticated over the length of the curve, and bone graft laid on.

Five days later, a pleural effusion developed. A chest drain was inserted; 150 ml of purulent blood-stained fluid was obtained and Staphylococcus aureus and Klebsiella aeruginosa were subsequently cultured. The patient improved on intravenous antibiotics over the next six days and the chest drain was removed. The following day she was unwell, cyanosed and hypoxic. Two chest drains were re-inserted, but 24 hours later septicemia developed and cardiac arrest occurred. She was successfully resuscitated but needed intermittent positive-pressure ventilation for 22 days. Large amounts of pus continued to discharge from the chest drain which remained in situ for a further nine days. She was discharged 51 days after the second procedure but could not tolerate any form of external support. Fifteen months later she is severely disabled because of breathlessness.

**Case 2.** A baby boy was diagnosed as having PWS at the age of three months, and a left thoracic scoliosis was noted at the age of seven months. Despite bracing, the curve measured 90° at the age of 5 years 8 months (Fig. 3). Pulmonary function testing was not possible since he could not co-operate; he could, however, run the length of the ward without difficulty. Because his parents would not consent to an anterior growth arrest as an initial procedure, surgery at the age of 6 years 3 months consisted of an uneventful posterior Luque instrumentation with segmental sublaminar wiring from T3 to L1 (Figs 4 and 5). Two weeks later he underwent an anterior growth arrest in which six discs and end plates were
excised from the apex of the curve and a rib was used as graft. A chest drain was inserted at this time and removed 48 hours later. Intravenous antibiotics were given prophylactically.

Recovery was uneventful until the fifth day when a pneumothorax developed, needing the re-insertion of a chest drain. Initially, a small amount of straw-coloured fluid was obtained. The drainage became profuse two days later and pseudomonas and coliform bacilli were subsequently cultured. On the same day meningism and drowsiness developed. Biochemical analysis suggested a leak of cerebrospinal fluid, and this was confirmed by myelography.

He remained seriously ill over the next two weeks, needing parenteral nutrition. The pleural drainage ceased spontaneously 30 days after the chest drain was reinserted. He wore a plaster jacket for nine months after discharge and is currently in a Plastazote spinal support with no loss of correction of his scoliosis.  

Case 3. A young child was diagnosed as having PWS at 10 years of age when she was referred to a paediatrician because of her obesity and short stature. She was also of low intelligence. A left thoracic scoliosis measuring 92° from T6 to L2 was noted (Fig. 6). All lung function tests were reduced to 50% of predicted values.

After successful dieting, a costectomy was performed at 11 years 1 month. The pleural cavity was not entered and recovery was uneventful. Two weeks later she underwent posterior Harrington rod instrumentation with fusion from T6 to L2. There was considerable bleeding (1750 ml) during surgery but the postoperative period was uneventful. At 15 months after operation, she was freed of all external support and the operative correction has been maintained. It is noteworthy that she sustained a femoral neck fracture at the age of 13 after a minor fall. The association of fractures following minor injury has previously been reported (Holm and Laurnen 1981).

DISCUSSION

Of the seven patients with PWS seen at Royal Liverpool Children’s Hospital, five are known to have spinal deformity. In addition to the three cases presented here, a 14-year-old boy with lumbar scoliosis measuring 13° and a 10-year-old boy with a 74° kyphosis are under review, thus giving a prevalence rate of 57% for scoliosis and 14% for kyphosis. Minor surgery does not seem to pose problems for these patients since four have had uneventful surgery for squints, herniae and undescended testicles.

There are few reports of surgery for scoliosis in PWS. Gurd and Thompson (1981) reported two girls: one was 13 years old, but no further details were given, and the other was seven years old with a 94° scoliosis from T6 to L2. After four months of skeletal traction, the seven-year-old child underwent surgery which was described as ‘unremarkable’ although the laminae were too weak to hold a Harrington rod. Holm and Laurnen (1981) reported a 12-year-old boy who underwent Harrington rod instrumentation and fusion for a 65° thoracic scoliosis. A correction to 30° was obtained. No details of the operation were recorded but convalescence was reported to be ‘uneventful’. Soriano et al (1988) reported a 16-year-old girl who had Harrington rod instrumentation and fusion for a 75° thoracic curve which was corrected to 45° uneventfully. All these cases had a single posterior procedure and no thoracotomy, as did one of our patients in whom surgery was uneventful apart from excessive bleeding.

Children with PWS are susceptible to respiratory problems for several reasons. Obesity and a short stature results in reduced respiratory function. Our two patients who could co-operate with pulmonary function tests had significantly impaired tidal volumes. Thoracotomy therefore presents a formidable challenge to their respiratory reserve, especially if ribs have been taped during wound closure. There is risk of chest infection. In addition, some authors have noted that myopathy may be part of PWS (Zellweger and Schneider 1968; Gurd and Thompson 1981) and may impair the function of respiratory muscles. Because of these patients’ impaired intellect, there is often difficulty in eliciting co-operation with postoperative chest physiotherapy.

Excessive bleeding at operation was a considerable problem in two of our patients. Although patients are
susceptible to bruising (Cassidy 1987), no coagulation defect has been detected. We have the impression that the tendency to bleed is due to a generalised friability of all tissues, suggesting a connective tissue disorder. Indeed, the strongest connective tissue of all, bone, is often too soft to provide adequate fixation of metalwork.

We therefore do not agree with Gurd and Thompson (1981), who stated that there do not seem to be any major problems in the surgical treatment of scoliosis in PWS. The frequency of serious postoperative complications in other children undergoing two-stage surgery in our unit is much less than that encountered in our PWS series. Major two-stage spinal surgery is often needed in scoliosis to achieve adequate correction and control of growth, especially in very young children, and posterior surgery alone may be inadequate. However, thoracotomy appears to have a high rate of formidable complications which need to be carefully considered; the risks must be fully appreciated by both surgeon and parents.

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REFERENCES


